chapter

18 The Basis of Heredity

In this chapter

Have you ever been able to identify a person as a member of a particular family by certain physical traits? Some traits, such as curly hair or a prominent nose, can be traced through a family's lineage. Heredity is the transmission of biological traits from parents to offspring. When the members of different generations all share a particular trait, this is evidence that the trait is inherited. Genetics is the study of inheritance of biological traits.

Biological traits are determined by genes, which are specific segments of DNA. During reproduction, genes of the parent or parents are transmitted to the next generation. Long before we knew of genes and DNA, humans were able to use knowledge of transmission of biological traits to their advantage. Domesticated animals, such as cows and dogs, were produced by choosing parents having traits that were desired in the offspring. Crop plants were also developed by selecting parents with desirable traits.

Every person inherits one of about eight million possible combinations of his or her parents' chromosomes. Your set of genes and your traits are therefore all your own. Even twins who are genetically identical may not share all the same traits.

What patterns can be found in the transmission of genetic traits? How do these relate to the transmission of genes? In this chapter, you will explore patterns of inheritance of biological traits and explain how these patterns arise.

STARTING Points

Answer these questions as best you can with your current knowledge. Then, using the concepts and skills you have learned, you will revise your answers at the end of the chapter.

1. Is it possible for two parents with black hair to have a child with red hair? Why or why not?
2. Sometimes, when breeders cross two individuals with valuable traits, the offspring do not show the same traits. Suggest a reason why this may be so.
3. A team of researchers at the University of Alberta studied sets of identical twins to see if driving a truck or other heavy machinery was related to back pain. Each set of twins included one individual who drove for a living and another who did not. They found that the amount of back pain experienced by a truck-driving twin was the same as for the non-driving twin.
   (a) Why was it important to study identical twins?
   (b) Could the study have used fraternal twins instead? Why or why not?

Career Connections:
Veterinarian; Agrologist

Figure 1
Bobby Hull and Brett Hull starred in the National Hockey League and were the first father and son to win the Hart Trophy.

Figure 2
The father of former prime minister Paul Martin was also a federal politician.

Figure 3
Keifer and Donald Sutherland have successful acting careers.

Exploration — Similarities and Differences

Look at the people shown in **Figure 1**, **Figure 2**, and **Figure 3**. Identify any traits, such as eye colour, eye shape, face shape, and nose length and width, that show a family resemblance. Consider the information in the captions.

- Organize the traits in a chart or table.
- Identify the traits that you think are inherited.

(a) Describe the criteria you used to decide that a trait was inherited.

(b) Brett Hull is one of the NHL's all-time goal scorers. Do you think Brett inherited the ability to score goals from his father, Bobby Hull (**Figure 1**), or is this a skill he learned? Give reasons for your answer.

18.1 Gregor Mendel—Pioneer of Genetics

Figure 1
Gregor Mendel (1822–1884) was an Austrian monk whose experiments with garden peas laid the foundation for the science of genetics.

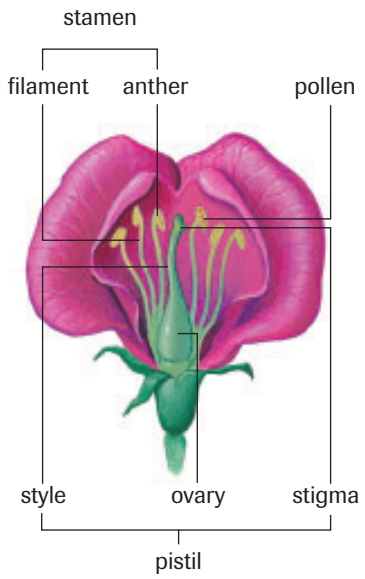

Figure 2
The structure of a flower

progeny new individuals that result from reproduction; offspring

dominant trait a characteristic that is expressed when one or both alleles in an individual are the dominant form

Humans have long understood that certain characteristics were passed down from generation to generation. Stone tablets crafted by the Babylonians 6000 years ago show pedigrees of successive generations of champion horses. However, the first real understanding of inheritance would not come until the work of an Austrian monk, Gregor Mendel, in the mid-19th century (**Figure 1**). Mendel tracked and recorded the transmission of seven visible traits through several generations of the garden pea. To keep track of the different generations, he called the first cross the parental generation, or P generation. The offspring of this cross he called the first filial generation, or the F_1 generation. The next generations were the F_2 generation, the F_3 generation, and so on.

Why did Mendel work with the garden pea? First, he observed that garden peas have a number of characteristics that are expressed in one of only two alternative forms. This made it easier to see which form was inherited.

The second reason is related to how this species reproduces. Garden peas usually reproduce through self-pollination. During pollination, the pollen produced by the anthers of the stamens attaches to the pistil. The pistil consists of the stigma, style, and ovary (**Figure 2**). The ovary contains an egg cell or female sex cell (gamete). Sperm cells (the male gametes) in the pollen grains fertilize the egg cell, and seeds are produced. In self-pollination, the pollen grains and the pistil are from the same plant: in cross-pollination, the pollen grains and the pistil are from different plants. The garden peas that Mendel worked with were "pure" varieties with known traits that came from a long line of self-pollinated pea plants. The traits of each variety had, therefore, been present in all individuals of that variety over many generations.

The Principle of Dominance

When Mendel used pollen from a pea plant with round seeds to fertilize a pea plant with wrinkled seeds, he found that all the offspring (the **progeny**) in the F_1 generation had round seeds. Did this mean that the pollen determines the shape of a seed? Mendel tested this by using pollen from a plant with wrinkled seeds to fertilize a plant with round seeds. Once again, all the progeny had round seeds. Round-seed shape was always the **dominant trait**, regardless of parentage. Mendel called the other wrinkled-seed shape the **recessive trait**. Mendel repeated the experiment for other traits. One trait was always dominant and the other recessive.

Mendel reasoned that each trait must be determined by something he called "factors." Today, we know these factors are genes. Mendel also realized that there can be alternate forms of a gene, which give rise to alternate forms of a trait. We now call the alternate form of a gene an **allele**. For example, the gene for seed colour has two alleles, one that determines green-seed colour and one that determines yellow. Alleles that determine dominant traits are dominant alleles. Alleles that determine recessive traits are recessive alleles. A dominant allele is indicated by an uppercase italic letter, such as *R* for round seeds. The recessive allele is designated by the lowercase italic letter, such as *r* for wrinkled seeds.

Mendel's Principle of Segregation

Mendel next let the F_1 plants self-fertilize, to observe the pattern of transmission of traits in the F_2 generation. When he had crossed pure round-seed plants with pure wrinkled-seed plants, 100 % of the F_1 generation had round seeds. Mendel was astonished to find that 75 % of the F_2 generation had round seeds and 25 % had wrinkled seeds. That is, for seed shape, the ratio was 3:1 round to wrinkled. He performed crosses to follow other traits and found the F_1 generations all had the same 3:1 ratio of dominant to recessive trait.

recessive trait a characteristic that is expressed only when both alleles in an individual are the recessive form

allele one of alternative forms of a gene

To explain these ratios, Mendel reasoned that each plant must carry two copies (alleles) of each gene that can be the same or different. An individual with round seeds must carry at least one dominant allele (*R*), but individuals with wrinkled seeds must always carry two copies of the recessive allele (*rr*).

When both alleles of a gene pair are the same, an individual is said to be **homozygous** for that trait. When the alleles of a gene pair are different, an individual is **heterozygous** for that trait. The complement of genes of an organism is called its **genotype**, and the physical expression of the genotype is the **phenotype**.

homozygous having identical alleles for the same gene

heterozygous having different alleles for the same gene

genotype the genetic complement of an organism

phenotype the observable characteristics of an organism

Mendel also correctly concluded that the two copies of a gene in a gene pair undergo **segregation** during the formation of the sex cells. Each mature gamete contains only one member of a gene pair. When an individual is homozygous for a gene, all of its gametes carry the same allele. When an individual is heterozygous for a gene, each gamete could receive either allele. **Figure 3 (a)** shows the results of a cross between two homozygous peas. At fertilization, the new individual receives one copy of the gene from the female parent and one from the male parent. All members of the F_1 generation, therefore, are heterozygous. When the F_1 generation was allowed to self-pollinate, three different genotypes were produced, which determined the two phenotypes that Mendel observed (**Figure 3 (b)**).

segregation the separation of alleles during meiosis

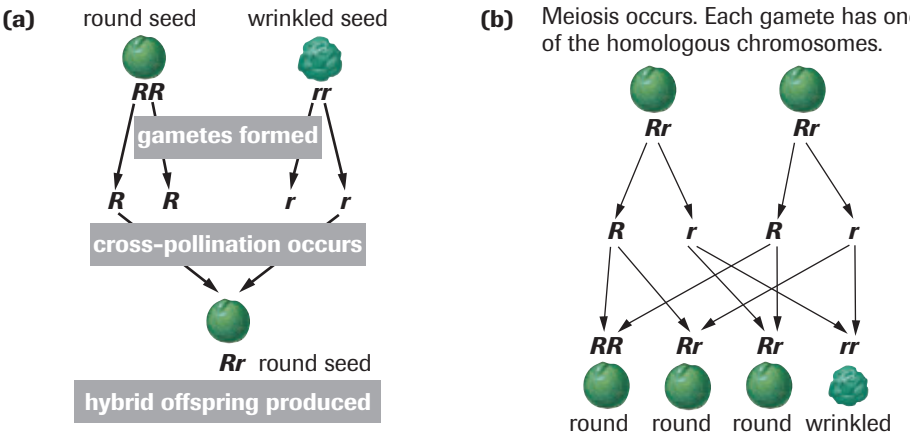

Figure 3

(a) When a pea plant homozygous for round seeds is cross-pollinated with a pea plant homozygous for wrinkled seeds, the offspring are all heterozygous.

(b) The F_2 progeny from a cross of two heterozygous pea plants with round seeds will have three possible genotypes *RR*, *Rr*, and *rr*.

EXTENSION

Genetic Terms

This animation gives a visual review of some of the terms used in studying genetics.

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mini Investigation Cross-Pollination

Materials: two plants of the same species that have different colours of flowers, small scissors, paint brush, plastic bags, potting soil, water, small pots

- On the plant you want to be the seed-parent, select a flower that is not yet open. Using **Figure 4** as a guide, remove the anthers from the flower.
- Using the paint brush, transfer pollen from the pollen-parent to the stigma of the seed-parent flower from which you removed the anthers.

(a) Predict the flower colour of the offspring of your cross-pollinated plant. Give reasons for your prediction.

(b) Why were the anthers removed from the plant that received the pollen?

(c) Why was a plastic bag placed over the flower?

- If there is time, collect and grow seeds from the flower you pollinated. Cover the pollinated flower with a plastic bag. Once the flower has produced seeds, plant the seeds in moist soil. Place the plant in sunlight (or under a bright light) and keep it watered until it produces flowers.

(d) Was your prediction correct?

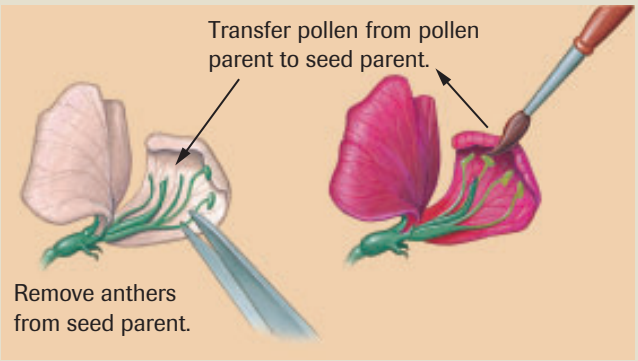

Figure 4
Pollen is transferred from the donor plant to the pistil of the recipient, which has had its stamens removed to prevent self-pollination.

SUMMARY Gregor Mendel—Pioneer of Genetics

- Inherited traits are controlled by factors—genes—that occur in pairs. Each member of a pair of genes is called an allele.
- One factor, or allele, masks the expression of another. This is known as the principle of dominance.
- A pair of factors, or alleles, separates from one another (segregate) during the formation of sex cells. This is often referred to as the law of segregation.

Section 18.1 Questions

1. Why were the pea plants selected by Mendel ideally suited for studying the transmission of traits?

2. Explain why, under normal circumstances, an individual can carry only two alleles of a gene.

3. Use an example that helps differentiate between the terms genotype and phenotype.

4. Black fur colour is dominant to yellow in Labrador retrievers.
(a) Explain how the genotype of a homozygous black dog differs from that of a heterozygous black dog.
(b) Could the heterozygous black dog have the same genotype as a yellow-haired dog? Explain.

5. A pea plant with round seeds is cross-pollinated with a pea plant that has wrinkled seeds. The plant with round seeds is heterozygous. Indicate each of the following:
(a) the genotypes of the parents
(b) the gametes produced by the parent with round seeds
(c) the gametes produced by the parent with wrinkled seeds
(d) the possible genotype(s) and the phenotype(s) of the F_1 generation

Probability and Inheritance of Single Traits 18.2

For every cross, Mendel kept track of the number of offspring that inherited the dominant trait and recessive trait. Based on mathematical analysis of these numbers, Mendel also concluded that each gamete produced by a heterozygous individual has an equal chance of getting either allele of a gene pair. Recall that when Mendel allowed peas that were heterozygous for the seed shape allele to self-pollinate, 75 % of the F_2 generation had the round-seed phenotype and 25 % had the wrinkled-seed phenotype. In other words, the **phenotypic ratio** of offspring with the dominant trait to offspring with the recessive trait was 3 to 1. To get this ratio, each sex cell must have had an equal probability of getting the *R* allele as the *r* allele during the process of segregation.

The probability of an outcome is a measure of the likelihood that the outcome will occur. Probability may be expressed as a fraction, a decimal, or a percentage. Probability (*P*) can be determined using the following formula:

$$P = \frac{\text{number of ways that a given outcome can occur}}{\text{total number of possible outcomes}}$$

For example, you might calculate the probability of getting heads when you toss a coin. There is only one way of tossing heads, so the numerator is 1. Since there are two possible outcomes in total, the denominator is 2. Therefore, the probability *P* of tossing heads is $\frac{1}{2}$, or 0.5, or 50 %.

A **Punnett square** is a chart that can help us to predict the phenotypes of the progeny of a cross between parents of known genotypes, or to deduce the genotypes of parents from the observed phenotypic ratio of their progeny. Punnett squares also allow us to determine the expected ratio of the genotypes (**genotypic ratio**) and the phenotypes for a cross, and to state the probability of that particular genotype or phenotype will occur in the progeny of a cross.

phenotypic ratio the ratio of offspring with a dominant trait to the alternative, recessive trait

Punnett square a chart used to determine the predicted outcome of a genetic cross

genotypic ratio the ratio of offspring with each possible allele combination from a particular cross

SAMPLE exercise 1

A breeder crosses a pea plant with wrinkled seeds and a pea plant with round seeds. She knows that the plant with round seeds is heterozygous for the gene for seed shape. The allele for round seeds (*R*) is dominant over the allele for wrinkled seeds (*r*). Determine the expected genotypic ratio and phenotypic ratio of the progeny.

Solution

Since *r* is the recessive allele, the genotype of the plant with wrinkled seeds must be *rr*. Since the plant with round seeds is heterozygous, its genotype must be *Rr*. The symbols for the alleles in the gametes are written across the top and along the left side of the Punnett square (**Figure 1**). Each cell is then filled in by entering one allele from the top of the square and a second allele from the side of the square.

Figure 2 shows a completed Punnett square for a cross between a heterozygous round-seed pea plant and a wrinkled-seed pea plant. Two of the four cells show the genotype *Rr* and two show *rr*. The expected genotypic ratio in the progeny of *Rr* to *rr* is, therefore, 1:1. Offspring with genotype *Rr* will have round seeds, and those with genotype *rr* will have wrinkled seeds.

Therefore, the phenotypic ratio is 1:1 ($\frac{1}{2}$ round and $\frac{1}{2}$ wrinkled).

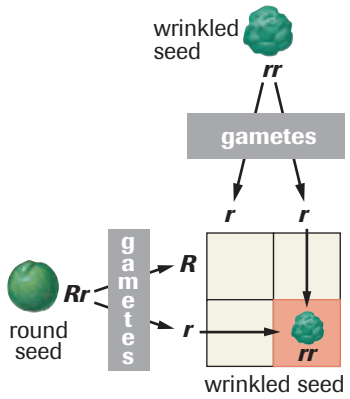

Figure 1
The partially completed Punnett square for a cross between a pea plant with genotype *rr* and a pea plant with genotype *Rr*. The genotype *rr* in one cell of the Punnett square is one of four possible combinations of the parental alleles.

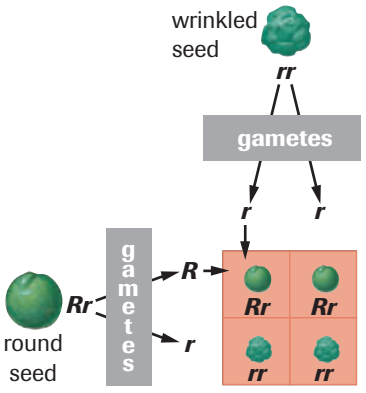

Figure 2

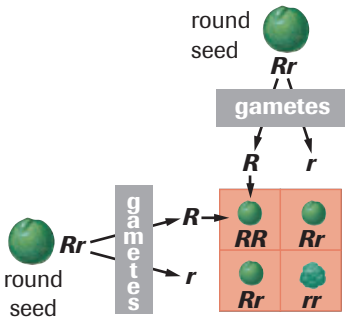

Figure 3
A Punnett square showing the results of a cross between two heterozygous plants with round seeds

SAMPLE exercise 2

For the cross shown in **Figure 3**, what is the probability that an offspring will have a phenotype of wrinkled seeds? Express the answer as a percent.

Solution

Since the allele for wrinkled seeds, *r*, is recessive, only offspring with a genotype *rr* will have wrinkled seeds. From the Punnett square, 1 of every 4 offspring are expected to have this genotype, so the probability that an offspring will have wrinkled seeds is 25 %.

Practice

1. What is the phenotypic ratio of the cross in the Punnett square shown in **Figure 4**?

	T	*T*
t	*Tt*	*Tt*
t	*Tt*	*Tt*

Figure 4
Punnett square of a monohybrid cross between a homozygous tall pea and a homozygous short pea

2. Using a Punnett square, determine the expected phenotypic ratio and genotypic ratio for the progeny of a cross between a pea plant that is homozygous for the white allele (*r*) for flower colour and a pea plant that is homozygous for the red allele (*R*).

EXTENSION

F_2 Ratios
This animation shows some of the results of Mendel's crosses, which you can then convert to phenotypic ratios. How close are the observed phenotypic ratios to the predicted phenotypic ratio?

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WEB Activity

Case Study—Creating a Personal Profile

Some human genes determine visible traits that show an inheritance pattern that is similar to that of Mendel's garden peas. As a result, you can predict a person's genotype for these traits just by observing him or her. In this activity, you will use a list of some common dominant and recessive traits, and use this information to create a profile of your own phenotype and potential genotype.

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EXTENSION

Genetics
In this Virtual Biology Laboratory, you can assess data and perform simulated crosses to explain the inheritance of shell colour in glyptodonts, an extinct relative of the armadillo.

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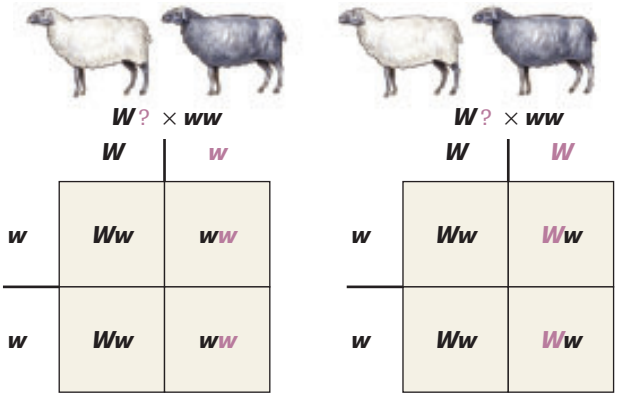
Test Crosses

Wool producers often prefer sheep with white wool, since black wool tends to be brittle and difficult to dye. Black sheep can be avoided by breeding only homozygous white rams. However, the allele for white wool *(W)* is dominant over the allele for black wool *(w)*, so white rams can be heterozygous. How could a wool producer be sure that a white ram is homozygous?

A **test cross** is the cross of an individual of unknown genotype to an individual with a recessive genotype. The phenotypes of the F_1 generation of a test cross reveal whether an individual with a dominant trait (such as a white ram) is homozygous or heterozygous for the dominant allele. If a white ram is crossed with a black ewe and the observed phenotypic ratio is 1:1 black to white, then the genotype of the ram must be *Ww* (**Figure 5**). If all the offspring are white, then the genotype of the white ram must be *WW*.

Test crosses are the simplest way of determining the genotype of an individual. Sometimes, however, the parents are not available to test cross. When only information about the phenotypes of the offspring of a cross is available, the genotypes and phenotypes of the parents can be found by working backwards through a Punnett square.

test cross the cross of an individual of unknown genotype to an individual that is fully recessive



Half of the offspring are black and half are white.

All of the offspring are white.

Figure 5
A test cross is a way of determining if an individual with the dominant trait is heterozygous or homozygous.

SAMPLE exercise 3

A horticulture worker has seeds from a particular cross, but has no information about the genotype or the phenotype of the parents. He plants and grows the offspring, and records the traits of each offspring **(Table 1)**. What was the genotype and phenotype of the parent plants?

Table 1

Offspring phenotype	Numbers
round-seed peas	5472
wrinkled-seed peas	1850

Solution

Determine the observed phenotypic ratio of the progeny, rounding off if needed.

$$\frac{\text{round}}{\text{wrinkled}} = \frac{5472}{1850} \approx \frac{3}{1}$$

List the possible genotypes for each phenotype, as shown in **Table 2**.

Table 2

Phenotype	Genotype
round-seed peas	*RR* or *Rr*
wrinkled-seed peas	*rr*

EXTENSION

Factors that Contribute to Genetic Variation

In this Audio Clip, you will hear about the underlying mechanisms that create genetic variation in the offspring of sexually reproducing individuals.

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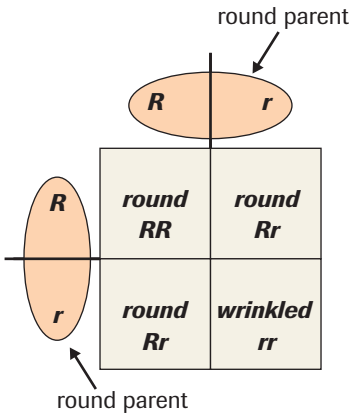

Figure 6
The observed phenotypic ratio is the same as the ratio predicted by the Punnett square.

A 3:1 phenotypic ratio occurs when two heterozygous individuals are crossed, so we know that the parents must be heterozygous. Since only $\frac{1}{4}$ of the progeny had wrinkled seeds, this is the recessive phenotype and must be determined by two copies of the recessive allele. The parents were heterozygous, so their genotype was *Rr*. Check the answer using a Punnett square **(Figure 6)**.

Practice

3. A fish breeder has a red male cichlid of unknown parentage. Red colour is dominant to yellow in the fish. He must know whether the fish is heterozygous for these colours. Suggest a way the fish breeder might find out the genotype of his red male. Use a Punnett square to explain your answer.

4. A neighbour gives a home gardener some seeds that he collected last year from his red carnations. The gardener plants 50 of the seeds and is surprised to find 12 of the plants have white flowers. Assuming that all the seeds came from one cross, what was the genotype of the parents?

SUMMARY Probability and Inheritance of Single Traits

- By using a Punnett square, the expected phenotypic ratio and genotypic ratio of the offspring of a cross can be determined.
- Probability, $P = \dfrac{\text{number of ways that a given outcome can occur}}{\text{total number of possible outcomes}}$

  Probability values can be used to predict the likelihood that a particular phenotype will appear in a cross.
- A test cross is the cross of an individual of unknown genotype to an individual with a fully recessive genotype.

Section 18.2 Questions

1. In Dalmatian dogs, the spotted condition is dominant to non-spotted.
   (a) Using a Punnett square, show the cross between two heterozygous parents.
   (b) A spotted female Dalmatian dog has six puppies sired by an unknown male. From their appearance, the owner concludes that the male was a Dalmatian. Three of the pups are spotted and three are not. What is the genotype and phenotype of the puppies' father?

2. For Mexican hairless dogs, the hairless trait is dominant to hairy. A litter of eight pups is found; six are hairless and two are hairy. What are the genotypes of their parents?

3. Test crosses are valuable tools for plant and animal breeders.
   (a) Provide two practical examples of why a cattle rancher might use a test cross.
   (b) Why are most test crosses performed using bulls rather than cows?

Pedigree Charts 18.3

Pedigree analysis is another tool for solving genetic problems. This approach is especially useful when it is not possible to perform crosses using specific individuals or to generate large numbers of progeny, such as for humans. A **pedigree chart** is like a family tree in which the inheritance of a trait can be traced from parents to offspring.

pedigree chart a chart used to record the transmission of a particular trait or traits over several generations

A pedigree chart shows the family relationship among individuals. Symbols identify the gender of each individual and whether an individual had the trait of interest. Pedigree charts may also show when an individual is known to be homozygous or heterozygous for a trait. The top of **Figure 1** shows some commonly used symbols. The pedigree chart in the lower part of **Figure 1** shows the transmission of an inherited disease among members of a family. Genetic counsellors may use pedigree charts in their work.

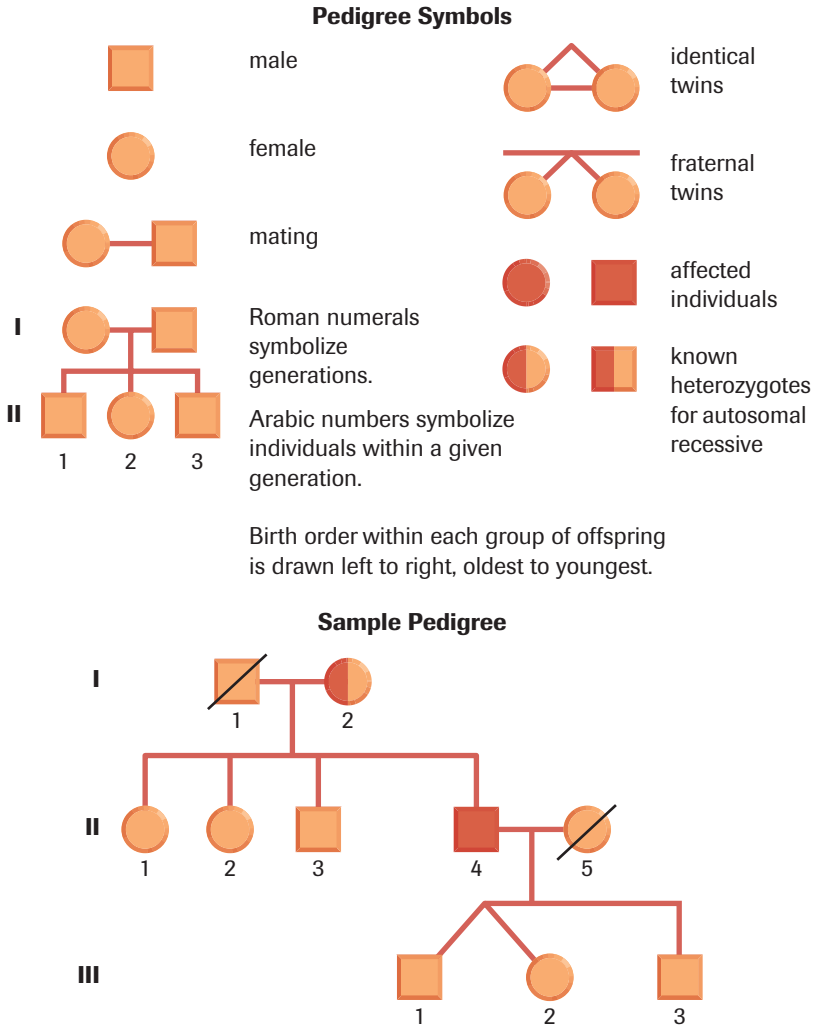

Figure 1
Squares represent males and circles represent females. A slash through a symbol indicates that person is deceased. Vertical lines connect parents to offspring, horizontal lines connect mates and connect siblings. Individuals affected by the inherited disease are identified by the darker-coloured symbols. Symbols having two different colours identify individuals heterozygous for the disease.

Practice

1. People with albinism do not produce normal pigment levels. Albinism is a recessive trait. Use the pedigree chart in **Figure 2** to answer the following questions. Use an uppercase "A" to represent the dominant allele, and a lowercase "a" for the recessive allele.
   (a) How many children do the parents A and B have?
   (b) Indicate the genotypes of the parents.
   (c) Give the genotypes of M and N.

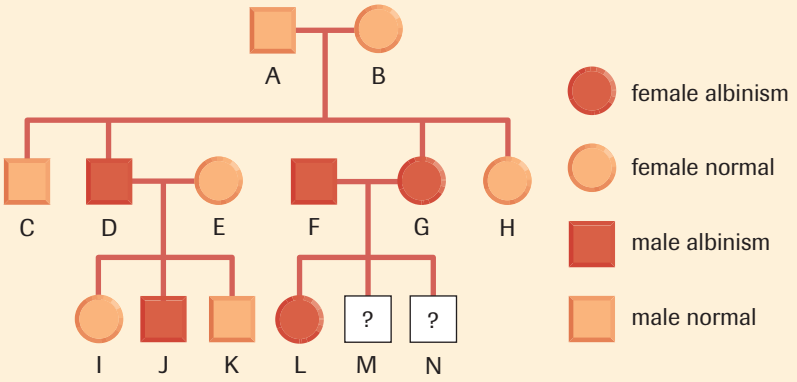

Figure 2

EXPLORE an issue

Genetic Screening

Due to advances in technology, it is now possible to get information about the genotype of any person relatively easily. Genetic screening may be carried out before birth (prenatal screening) or any time after birth. The most common reason for parents to want prenatal genetic screening is because they are at increased risk of passing a genetic disease to their child.

Thalassemia is one genetic disease for which prenatal genetic screening may be performed. Thalassemia is a disease of the blood, which affects a person's ability to produce enough red blood cells. Only people with two copies of a mutant allele of a particular a gene will have the disease. Genetic screening for thalassemia is performed only on those people with a family history of the disease. Prenatal screening can identify the presence of the thalassemia allele before the child is born.

Persons at risk of Huntington disease may request either pre- or post-natal screening. Huntington disease is a neurological disorder caused by a dominant allele. Huntington is characterized by rapid deterioration of nerve control, which causes a range of symptoms, including involuntary movements, slurred speech, loss of memory, and depression. Huntington disease is fatal. There is no cure and available treatments have little effect on symptoms. Symptoms of Huntington disease begin gradually, usually in middle age, when most people have already had children. Genetic screening allows people to know whether they have inherited the disease before any symptoms develop, so they may know whether they are at risk of passing it on to their children.

Issue Checklist

● Issue	○ Design	● Analysis
● Resolution	● Evidence	● Evaluation

Understanding the Issue

- Working in a group, conduct research and find out more about genetic screening.

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1. Define genetic screening. Describe some technologies used in genetic screening.
2. What are some advantages of genetic screening? Provide an example.
3. What are some physical dangers associated with genetic screening methods? Provide an example.

Take a Stand

Consider this position statement: Genetic screening should be compulsory for any person with a family history of a genetic disease.

With your group members, create a list of different stakeholders in this issue. Based on your research, determine points that support and refute the position statement from the perspective of each stakeholder. Then, decide whether your group agrees or disagrees with the position statement. Present your position to the class. Prepare to defend your group's position in a class discussion.

WEB Activity

Simulation—Pedigree Analysis

Complete the interactive Pedigree Analysis Tutorial in this Virtual Biology Laboratory. You can also use pedigree analysis to examine the inheritance of several genetic diseases in humans, and to act as a "genetic counsellor" in some hypothetical case studies.

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SUMMARY Pedigree Charts

- A pedigree chart traces the inheritance of a trait from parents to offspring through several generations.
- Pedigree charts are useful in cases when it is not possible to perform and follow specific crosses, such as in human genetic studies.

Section 18.3 Questions

1. A woman begins to show symptoms of Huntington disease. Her father had Huntington disease, but her mother never developed the disorder. Neither her husband nor anyone in his immediate family have any symptoms.
(a) What is the genotype of the woman with Huntington disease?
(b) What is the probable genotype of the woman's husband?
(c) If the woman has six children, how many are likely to develop Huntington disease?

2. Phenylketonuria (PKU) is a genetic disorder caused by a dominant allele. Individuals with PKU are unable to metabolize a naturally occurring amino acid, phenylalanine. If phenylalanine accumulates, it inhibits the development of the nervous system, leading to mental retardation. The symptoms of PKU are not usually evident at birth, but can develop quickly if the child is not placed on a special diet. The pedigree in **Figure 3** shows the inheritance of the defective PKU allele in a family.
(a) How many generations are shown by the pedigree?
(b) How many children were born to the parents of the first generation?

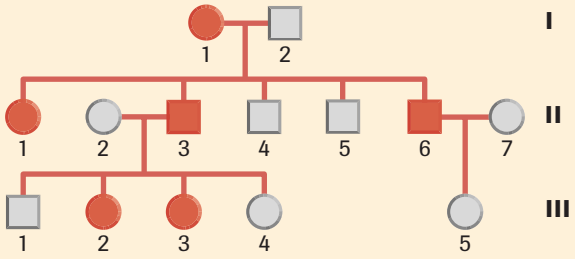

Figure 3

(c) What is the genotype of individuals 1 and 2, generation I?
(d) How is it possible that in generation II, some of the children showed symptoms of PKU, while others did not? (Hint: Use a Punnett square to help with your explanation.)
(e) For individuals 6 and 7, in generation II, a child without PKU symptoms was born. Does this mean that they can never have a child with PKU? Explain your answer.

3. Research the inheritance of one of the traits in **Table 1** in a family that you know. Get information from at least three generations of the family. Use the information you collect to make a pedigree chart.

Table 1

Trait	Dominant	Recessive
freckles	present	absent
dimples	present	absent
earlobe	suspended	attached
hairline	pointed on forehead	straight across forehead
chin dimple	present	absent

4. (a) How or where might genetic screening be used for purposes other than genetic counselling?
(b) What laws, if any, do you think are likely to arise regarding the use of genetic screening? Why?

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18.4 Other Patterns of Inheritance

pleiotropic gene a gene that affects more than one characteristic

EXTENSION

Pleiotropic Effects of Marfan Syndrome

Marfan Syndrome is caused by a mutation in a single gene. This animation shows you how this one gene affects four different organ systems.

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wild type the most common allele of a gene with multiple alleles

mutant any allele of a gene other than the wild type allele

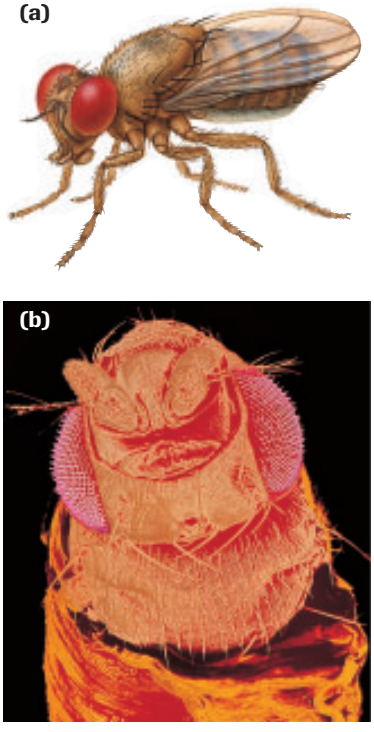

Figure 1

(a) *Drosophila melanogaster*, the fruit fly, is widely used for genetic studies.

(b) Wild type, or red, is the most common eye colour. It is dominant over all the other alleles for eye colour.

The traits that Mendel studied showed little variability. Each had only two alleles, one that was clearly dominant and one clearly recessive. However, many inherited traits show more variability than just two alternate forms. These types of traits will not be inherited in the predicted 3:1 phenotypic ratio of a trait with one dominant allele and one recessive allele.

Pleiotropic Genes

Some genes, called **pleiotropic genes**, affect many different characteristics. Sickle-cell anemia, a blood disorder, is caused by a pleiotropic gene. Normal hemoglobin (the pigment that carries oxygen in the blood) is produced by the allele *HbA*. Sickle cell anemia occurs in individuals who have two copies of the mutated allele, *HbS*. This mutation produces abnormally shaped hemoglobin molecules that interlock with one another. The new arrangement of molecules changes the shape of the red blood cells, which become bent into a sickle shape. The sickle-shaped red blood cells cannot pass through the capillaries, and so cannot deliver oxygen to the cells. People with sickle-cell anemia can suffer from fatigue and weakness, an enlarged spleen, rheumatism, and pneumonia. Patients often show signs of heart, kidney, lung, and muscle damage.

Multiple Alleles

When traits are determined by more than two (multiple) alleles, the most commonly seen trait is called the **wild type**, and the allele that determines it is the wild-type allele. Non-wild-type traits are said to be **mutant**, and the alleles that determine them are mutant alleles. In most cases of multiple alleles, there is a hierarchy of dominance.

Members of the species *Drosophila melanogaster*, the fruit fly (**Figure 1**), can have any one of four eye colours. Red eye colour is the wild type, but the eyes may also be apricot, honey, or white. The *Drosophila* species as a whole has more than two alleles for eye colour but, since fruit flies are diploid, each individual carries only two genes for eye colour.

The dominance hierarchy and symbols for eye colour in *Drosophila* are shown in **Table 1**. When there are multiple alleles for the same gene, upper case letters and superscript numbers are used to express the dominance relationships between the different alleles. For simplicity, the capital letter *E* is used for the eye colour gene and superscript numbers to indicate the position of each allele in the dominance hierarchy.

Table 1 Dominance Hierarchy and Symbols for Eye Colour in *Drosophila*

Phenotype	Allele symbol	Possible genotype(s)	Dominant over
wild type	E^1	E^1E^1, E^1E^2, E^1E^3, E^1E^4	apricot, honey, white
apricot	E^2	E^2E^2, E^2E^3, E^2E^4	honey, white
honey	E^3	E^3E^3, E^3E^4	white
white	E^4	E^4E^4	

SAMPLE exercise 1

What will be the phenotypic ratio of the offspring from the mating of the following *Drosophila* individuals?

E^1E^4 (wild-type eye colour) × E^2E^3 (apricot eye colour)

Solution

The problem can be solved by using a Punnett square. The first parent is heterozygous, and so will produce gametes with the E^1 allele and the E^4 allele. The other parent is also heterozygous, and will produce gametes with the E^2 allele and the E^3 allele. The Punnett square for this cross is, therefore, as shown in **Figure 2**.

Using the dominance hierarchy in **Table 1**, the phenotypic ratio of the F_1 offspring will produce two wild-type eye colour (genotypes E^1E^2 and E^1E^3) to one apricot eye colour (genotype E^2E^4) to one honey eye colour (genotype E^3E^4).

Practice

1. A student working with *Drosophila* makes the following cross:

   E^1E^2 (wild-type eye colour) × E^2E^4 (apricot eye colour)

   What will be the phenotypic ratio of the offspring?

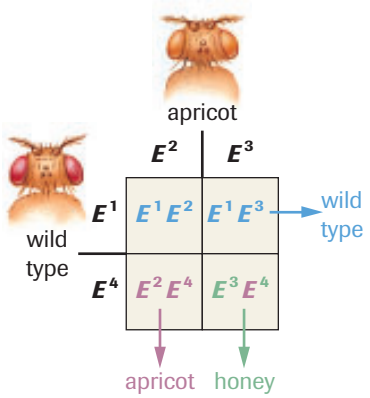

Figure 2
A cross between a fruit fly with wild-type eye colour and one with apricot-coloured eyes

Incomplete Dominance

When two alleles are equally dominant, they interact to produce a new phenotype—this form of interaction between alleles is known as **incomplete dominance**. When an individual is heterozygous for two alleles that show incomplete dominance, both alleles are equally expressed, but at half the level that would occur were the individual homozygous for either allele. The phenotype of a heterozygous individual is, therefore, intermediate between its homozygous parents. For example, when a homozygous red snapdragon is crossed with a homozygous white snapdragon, all of the F_1 generation have pink flowers. If members of the F_1 generation are crossed, the F_2 generation has a surprising phenotypic ratio of one red to two pink to one white (1:2:1). The Punnett square in **Figure 3** shows the genotypes behind this ratio.

incomplete dominance the expression of both forms of an allele in a heterozygous individual in the cells of an organism, producing an intermediate phenotype

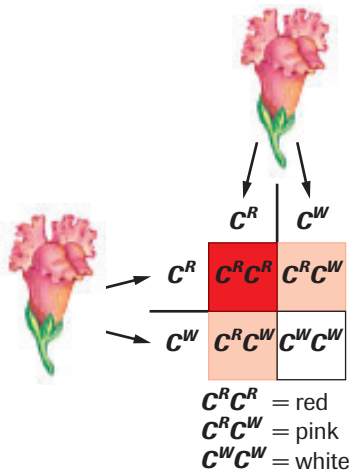

Figure 3
Colour in snapdragons is an example of incomplete dominance. When homozygous red-flowered snapdragons are crossed with homozygous white-flowered snapdragons, the F_1 generation all have pink flowers. When a cross is made between two F_1 individuals, the F_2 generation has a phenotypic ratio of one red to two pink to one white.

Codominance

codominance the expression of both forms of an allele in a heterozygous individual in different cells of the same organism

Another form of allele interaction is **codominance**. When two alleles show codominance, both alleles are fully expressed in a heterozygous individual, but not in the same cells. Coat colour in shorthorn cattle shows codominance (**Figure 4**). Red coats are composed of all red hairs, and white coats are all white hairs. When a red shorthorn is crossed with a white shorthorn, any calves produced will have roan-coloured coats, which is intermediate between the red and the white coat colour. However, each hair is not the intermediate roan colour. Instead, a roan coat has a mixture of white hairs and red hairs.

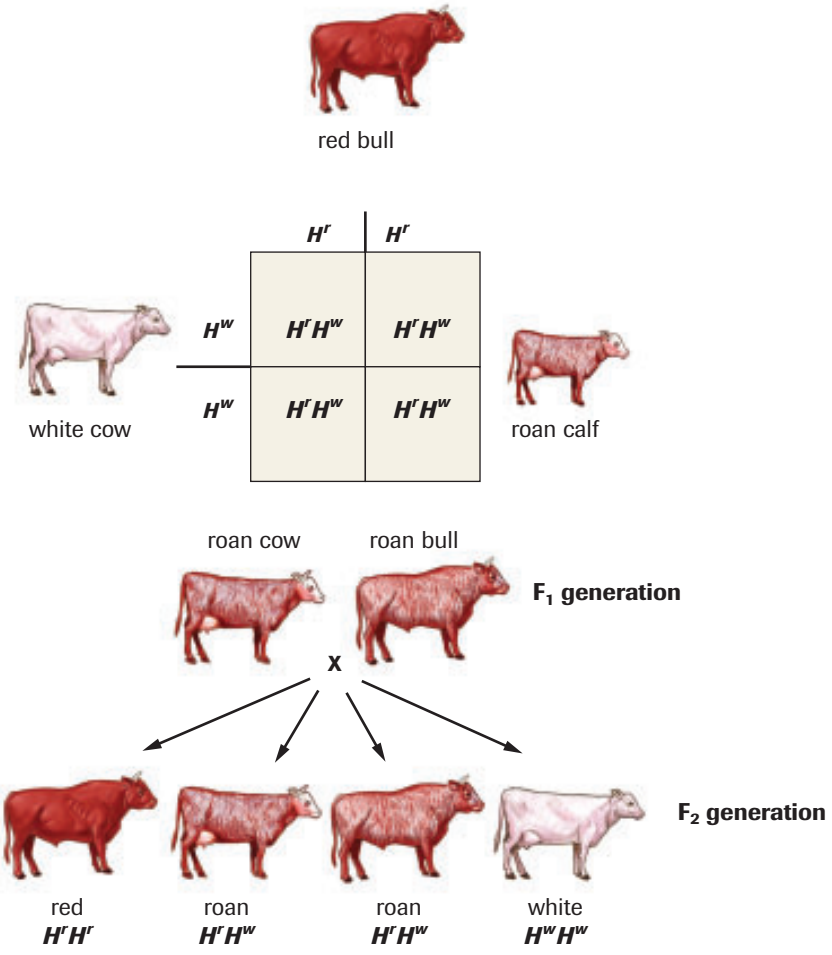

Figure 4
In codominance, either one of two different alleles is expressed. In shorthorn cattle, the coats of roan animals have intermingled red and white hair.

CAREER CONNECTION

Veterinarian
Veterinarians provide health care services that include the diagnosis and treatment of injured and sick animals. They give advice about the breeding of animals and perform genetic procedures and embryo transfers. Veterinarians work long hours and are dedicated animal health specialists. Learn more about their duties.

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Environment and Phenotype

Sometimes, variation of a trait is determined by the interaction of the genotype with the environment. The environment can have a profound effect on phenotype. Himalayan rabbits have black fur when they are raised at low temperatures, but white fur when raised at high temperatures. In some cases, different parts of the same organism can have different traits when exposed to different environments. Leaves of the water buttercup, *Ranunculus aquatilis*, that develop above the surface of the water are broad, lobed, and flat, while those that develop below the water are thin and finely divided. However, the leaves all have identical genetic information.

EXTENSION

Coat Colour in the Himalayan Rabbit
View this animation of how coat colour in this species is affected by temperature.

www.science.nelson.com GO

INVESTIGATION 18.1 *Introduction*

How Do Environmental Factors Affect Gene Expression?

Design and carry out an investigation of the effect of an environmental factor on the phenotype of genetically identical plants.

To perform this investigation, turn to page 620.

Report Checklist

- Purpose
- Problem
- Hypothesis
- Prediction
- Design
- Materials
- Procedure
- Evidence
- Analysis
- Evaluation
- Synthesis

Case Study

A Mystery of Blood Types

Humans have four blood types; A, B, AB, and O. The alleles for blood types A and B are codominant but dominant to O (**Table 2**). We also each have one of two forms of rhesus factor—the positive form (Rh+) or the negative form (Rh−). The allele for the Rh+ form is dominant to the Rh− allele. Blood types can identify individuals and family members.

Table 2 Human Blood Types

Phenotypes	Genotypes
Type A	I^AI^A, I^Ai
Type B	I^BI^B, I^Bi
Type AB	I^AI^B
Type O	ii

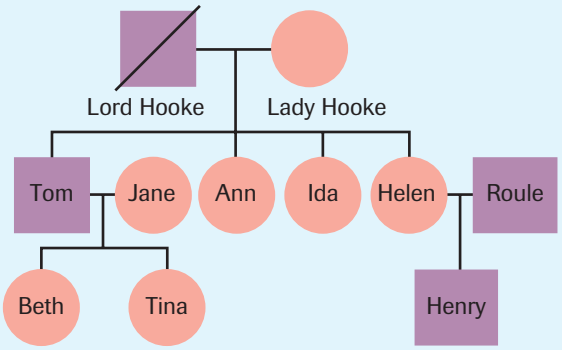


Figure 6
The family tree of the members of Lord Hooke's family who were in the castle

Evidence

In Black Mourning Castle, a scream echoed from the den of Lord Hooke. When the maid peered through the door, a freckled arm reached for her neck. She bolted and telephoned the police. Inspector Holmes arrived to find the dead body of Lord Hooke. Apparently, the lord had been strangled. The inspector noted blood on a letter opener, even though Lord Hooke did not have any cuts. This blood was type O, Rh−. Inspector Holmes took blood samples from the family members shown in **Figure 6**.

The inspector gathered the information shown in **Table 3**. The gene for freckles is dominant to the gene for no freckles. Some family members were wearing long-sleeved shirts, so the inspector could not determine whether freckles were present.

The inspector then announced, "Lady Hooke had been unfaithful to her husband. One of the heirs to the fortune was not Lord Hooke's child. The murder was committed to preserve a share of the fortune!"

Table 3 Traits of the Hooke Family

Family	Blood type	Rh factor	Freckles
Lord Hooke	AB	+	no
Lady Hooke	A	+	no
Helen	A	+	no
Roule	O	+	no
Henry	refused blood test		?
Ida	A	–	?
Ann	B	+	?
Tom	O	–	no
Jane	A	+	?
Beth	O	–	?
Tina	A	+	yes

Case Study Questions

1. Who was the murderer? What was the murderer's probable blood type?
2. Describe how you obtained your answer.
3. How did the inspector eliminate the other family members?

SUMMARY Other Patterns of Inheritance

- Some genes have more than two alleles, and can determine more than two forms of a trait. Multiple alleles may display a dominance hierarchy.
- Alleles that show incomplete dominance are equally dominant. An individual who is heterozygous for alleles that show incomplete dominance will have an intermediate phenotype.
- Codominant alleles are both expressed in a heterozygous individual.

Section 18.4 Questions

1. Multiple alleles control the coat colour of rabbits. A grey colour is produced by the dominant allele *C*. The C^{ch} allele produces a silver-grey colour, called chinchilla, when present in the homozygous condition, $C^{ch}C^{ch}$. When C^{ch} is present with a recessive gene, a light silver-grey colour is produced. The allele C^h is recessive to both the full-colour allele and the chinchilla allele. The C^h allele produces a white colour with black extremities. This coloration pattern is called Himalayan. An allele C^a is recessive to all genes. The C^a allele results in a lack of pigment, called albino. The dominance hierarchy is $C > C^{ch} > C^h > C^a$. **Table 4** provides the possible genotypes and phenotypes for coat colour in rabbits. Notice that four genotypes are possible for full-colour but only one for albino.

Table 4 Coat Colour in Himalayan Rabbits

Phenotypes	Genotypes
full colour	CC, CC^{ch}, CC^h, CC^a
chinchilla	$C^{ch}C^{ch}$
light grey	$C^{ch}C^h$, $C^{ch}C^a$
Himalayan	C^hC^h, C^hC^a
albino	C^aC^a

(a) Indicate the genotypes and phenotypes of the F_1 generation from the mating of a heterozygous Himalayan-coat rabbit with an albino-coat rabbit.
(b) The mating of a full-colour rabbit with a light-grey rabbit produces two full-colour offspring, one light-grey offspring, and one albino offspring. Indicate the genotypes of the parents.
(c) A chinchilla rabbit is mated with a light-grey rabbit. The breeder knows that the light-grey rabbit had an albino mother. Indicate the genotypes and phenotypes of the F_1 generation from this mating.
(d) A test cross is performed with a light-grey rabbit, and the following offspring are noted: five Himalayan rabbits and five light-grey rabbits. Indicate the genotype of the light-grey rabbit.

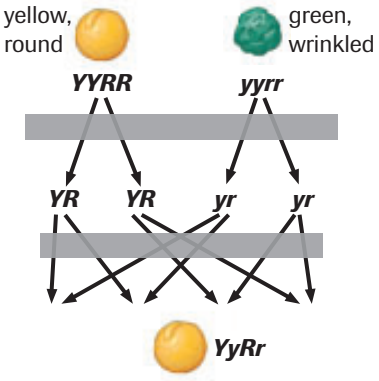
2. A horse that is homozygous for the allele C^r will have a chestnut, or reddish, coat. A horse that is homozygous for the allele C^m will have a very pale cream coat, called cremello. Palomino coat colour is determined by the interaction of both the chestnut and the cremello allele. Indicate the expected genotypic ratio and phenotypic ratio of the F_1 progeny of a palomino horse with a cremello horse.

3. Two pea plants are cross-pollinated. Using a Punnett square and probability analysis, you predict that $\frac{3}{4}$ of the offspring will be tall. However, less than $\frac{1}{4}$ grow to be tall. What other factors can affect phenotype? How much trust should be put on probability calculations?

Dihybrid Crosses and Polygenic Traits 18.5

A **dihybrid cross** is a cross that involves individuals with two independent traits that are present in alternate forms. Mendel performed dihybrid crosses with his garden peas to see if traits were inherited independently or with one another. He first crossed plants that were pure-breeding (homozygous) for two dominant traits with plants that were homozygous for two recessive traits, as shown in **Figure 1**. Each parent is homozygous for two traits, seed shape and seed colour. All the members of the F_1 offspring are heterozygous for the seed-colour gene and for the seed-shape gene. Since all the F_1 progeny had yellow, round seeds, Mendel's principle of dominance applies to this dihybrid cross.

dihybrid cross a genetic cross involving two genes, each of which has more than one allele



All members of the F_1 generation have the same genotype and phenotype.

Figure 1
A dihybrid cross between a pea plant that is homozygous for yellow seed colour (*YY*) and round seed shape (*RR*) with a plant that is homozygous for green seed colour (*yy*) and wrinkled seed shape (*rr*).

Evidence of Independent Assortment

Mendel explained the result shown in **Figure 1** by postulating that each gene was inherited independently. Today, this is referred to as Mendel's second law or the law of *independent assortment*. This law states that genes that are located on different chromosomes assort independently.

To create a Punnett square for a dihybrid cross, we include one allele for both of the genes in the possible gametes. The Punnett square in **Figure 2** shows the expected genotypes and phenotypes for Mendel's dihybrid cross when we assume that the genes for seed shape and seed colour are inherited independently. One parent will produce gametes with alleles *yR* and the other will produce gametes with alleles *Yr*. The predicted phenotype of the F_1 generation is the same as Mendel observed.

Figure 3 shows the behaviour of two separate chromosomes, one that carries the gene for seed shape and another that carries the gene for seed colour. (Pea plants actually have more than two chromosomes.) As the homologous chromosomes move to opposite poles during meiosis, each gamete receives two chromosomes, one carrying the seed-shape gene and one carrying the seed-colour gene. According to the law of segregation, the alleles of both these genes will segregate during meiosis. Therefore, the allele for yellow seeds segregates from the allele for round seeds, and the allele for wrinkled seeds segregates from the allele for round seeds.

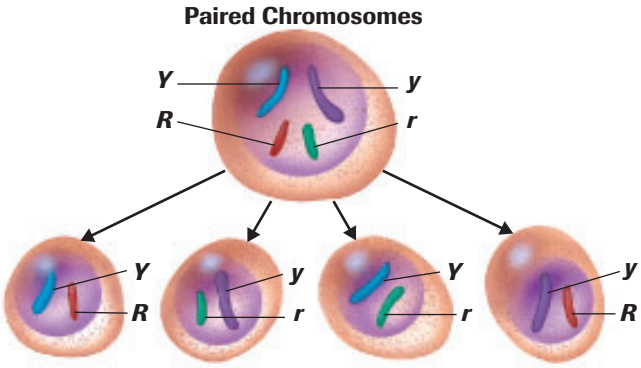


Figure 3
Segregation of alleles and independent assortment of chromosomes during meiosis gives rise to four possible combinations of alleles in the gametes of a plant of genotype *YyRr*.

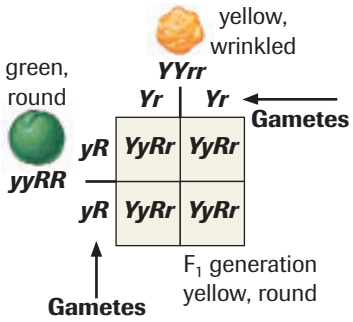


Figure 2
All gametes produced by a pea plant homozygous for yellow seed colour (*YY*) and wrinkled seed shape (*rr*) will have the alleles *Yr*. Similarly, all gametes produced by a pea plant homozygous for green seed colour (*yy*) and round seed shape (*RR*) will have the alleles *yR*. Since all the offspring have yellow, round seeds, the genotype of all the F_1 generation must be *YyRr*. This would not be possible if the genes for seed shape and seed colour were inherited together.

Mendel then produced an F_2 generation by allowing the F_1 progeny to self-fertilize. He recorded the phenotypes of all the F_2 progeny and then calculated the ratio of each phenotype he observed. The F_2 generation had the following phenotypic ratios: $\frac{9}{16}$ yellow, round seeds; $\frac{3}{16}$ green, round seeds; $\frac{3}{16}$ yellow, wrinkled seeds; and $\frac{1}{16}$ green, wrinkled seeds.

Figure 4 shows the expected genotypes from this cross when we assume that independent assortment occurred. The parents would produce four types of gametes. The genotypes in nine of the 16 cells would determine yellow, round seeds (*YYRR*, *YyRR*, *YYRr*, and *YyRr*); three of the 16 cells would determine green, round seeds (*yyRR* and *yyRr*); three more cells would determine yellow, wrinkled seeds (*YYrr* and *Yyrr*); and one cell would determine green, wrinkled seeds (*yyrr*). Since the predicted phenotypic ratio is the same as the ratio that Mendel observed, this cross also provides evidence for independent assortment.

Gametes	*YR*	*yR*	*Yr*	*yr*
YR	*YYRR*	*YyRR*	*YYRr*	*YyRr*
yR	*YyRR*	*yyRR*	*YyRr*	*yyRr*
Yr	*YYRr*	*YyRr*	*YYrr*	*Yyrr*
yr	*YyRr*	*yyRr*	*Yyrr*	*yyrr*

Figure 4
From the Punnett square analysis, self-fertilization of the F_1 generation will result in an F_2 generation with a 9:3:3:1 ratio. This ratio can only result if segregation of alleles and independent assortment of chromosomes occurs.

INVESTIGATION 18.2 ***Introduction***

Genetics of Corn

Use Punnett squares and phenotypic ratios to analyze the inheritance of two traits in corn.

Report Checklist

○ Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

To perform this investigation, turn to page 620.

Probability and Dihybrid Crosses

We can determine the probability of particular phenotypes and genotypes in the progeny of dihybrid crosses in much the same way as for monohybrid crosses. Probability values can be used to predict the chances of getting a particular genotype or phenotype in an offspring, or to tell us whether two genes are likely to be located on different chromosomes. In dihybrid crosses, however, we are interested in finding out the probability that two outcomes will occur at the same time. Recall that probability (P) is given by

$$P = \frac{\text{number of ways that a given outcome can occur}}{\text{total number of possible outcomes}}$$

SAMPLE exercise 1

In humans, free ear lobes are determined by the dominant allele *E*, and attached ear lobes by the recessive allele *e*. The dominant allele *W* determines a widow's peak hairline and the recessive allele *w* determines a straight hairline (**Figure 5**). The genes for these two traits are located on different chromosomes. Suppose a man with the genotype *EeWw* and a woman with the genotype *EeWw* are expecting a child. What is the probability that the child will have a straight hairline and attached ear lobes?

(a)

(b)

(c)

(d)

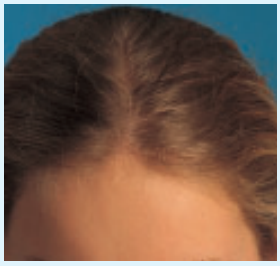


Figure 5
In humans, both ear lobe shape and hairline shape are inherited. The free ear lobe in **(a)** is dominant to the attached ear lobe in **(b)**, and the widow's peak in **(c)** is dominant to a straight hairline in **(d)**.

Solution

To have attached ear lobes and a straight hairline, the child must have the genotype *eeww*. Since the two genes are on separate chromosomes, the gene for ear shape and hairline shape will assort independently. The outcome that the child will receive two *e* alleles is, therefore, independent of the outcome that the child will receive two *w* alleles.

First, determine the probability of each of these outcomes separately, using a separate Punnett square for each gene. From **Figure 6 (a)**, we see the probability that the child will have attached ear lobes is one in four ($\frac{1}{4}$). From **Figure 6 (b)**, we see the probability that the child will have a straight hairline is also one in four ($\frac{1}{4}$).

(a)

	$\frac{1}{2}$ *E*	$\frac{1}{2}$ *e*
$\frac{1}{2}$ *E*	$\frac{1}{4}$ *EE*	$\frac{1}{4}$ *Ee*
$\frac{1}{2}$ *e*	$\frac{1}{4}$ *Ee*	$\frac{1}{4}$ *ee*

(b)

	$\frac{1}{2}$ *W*	$\frac{1}{2}$ *w*
$\frac{1}{2}$ *W*	$\frac{1}{4}$ *WW*	$\frac{1}{4}$ *Ww*
$\frac{1}{2}$ *w*	$\frac{1}{4}$ *Ww*	$\frac{1}{4}$ *ww*

Figure 6
Punnett squares showing monohybrid crosses between heterozygous parents for **(a)** free ear lobes and **(b)** for a widow's peak

Learning Tip

When thinking about probability, keep the following two rules in mind:

- When outcomes are independent, the probability of one outcome is not affected by the result of any other outcomes. For example, if you toss two heads in a row, the probability of tossing heads a third time is still 1 out of 2.
- The probability of independent events occurring together is equal to the *product* of those events occurring separately. The chances of tossing heads once is $\frac{1}{2}$, the probability of tossing heads twice in a row is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$, and the probability of tossing heads three times in a row is $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$.

EXTENSION

Probability—The Sum and Product Rules

This Audio Clip explores the use of the sum and product rules of probability.

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CAREER CONNECTION

Agrologist

Agrologists are plant, crop, and food production specialists. New breeds of plants and animals are of great interest to these scientists. They work with grain farmers and livestock producers on research projects designed to overcome challenges and realize economic opportunities in agriculture. Learn how agrologists specialize in many fields.

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Now, multiply these probabilities to calculate the probabilities of each event occurring in a dihybrid cross—that is, for the combination of traits. Therefore, the probability that the child will have the genotype *eeww* is $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$.

Practice

1. Calculate the probability that the couple will have a child with
   (a) a widow's peak and free ear lobes
   (b) a straight hairline and free ear lobes
   (c) a widow's peak and attached ear lobes

Selective Breeding

selective breeding the crossing of desired traits from plants or animals to produce offspring with both characteristics

The plants and animals that make up the world's food supply have, in large part, been developed artificially from wild ancestors. **Selective breeding** involves identifying individuals with desirable traits and using them as parents for the next generation. Over time, the desirable traits became more and more common. For example, North American Aboriginal farmers used selective breeding to develop many useful crop plants, long before the arrival of Europeans. Many crops that are important to Canadian agriculture were developed by selective breeding, including rust-resistant wheat; sweet, full-kernel corn; and canola, which germinates and grows rapidly in colder climates.

inbreeding the process whereby breeding stock is drawn from a limited number of individuals possessing desirable phenotypes

You are probably familiar with the term "purebreds." Many dogs and horses are considered to be purebreds, or thoroughbreds. Genotypes of these animals are closely regulated by a process called **inbreeding,** in which similar phenotypes are selected for breeding. The desirable traits vary from breed to breed. For example, Irish setters are bred for their long, narrow facial structure and long, wispy hair, but dalmations are bred for broader faces and short hair with spots. The bull terrier (pit bull) was originally bred for fighting. Quick reflexes and strong jaws were chosen as desirable phenotypes. Some geneticists have complained that inbreeding has caused problems for the general public as well as for the breed itself.

DID YOU KNOW

Aboriginal Crop Plants

For centuries, Aboriginal peoples bred many crop plants besides corn, which they ultimately introduced to European settlers. These include beans, tomatoes, potatoes, peanuts, peppers, cocoa, squash, pumpkins, sunflowers, long-fibre cotton, rubber, and quinine.

New varieties of plants and animals can be developed by hybridization. This process is the opposite to that of inbreeding. Rather than breed plants or animals with similar traits, the hybridization technique attempts to blend desirable but different traits. Corn has been hybridized extensively, beginning with the work of Aboriginal peoples thousands of years ago. The hybrids tend to be more vigorous than either parent. **Figure 7**, on the next page, shows the most common method used. Two homozygous plants, A and B, are crossed to produce an AB hybrid. Two other homozygous plants, C and D, are crossed to produce a CD hybrid. Hybrids AB and CD are then crossed to produce hybrid ABCD. This hybrid will have desired traits from plants A, B, C, and D, and will be more vigorous.

Polygenic Traits

polygenic trait inherited characteristics that are determined by more than one gene

In dihybrid crosses, two genes determine two separate traits. However, sometimes a single trait is determined by more than one gene. Many of your characteristics are determined by several pairs of independent genes. Skin colour, eye color, and height are but a few of your characteristics that are **polygenic traits**. Polygenic traits have much more variability in a population than those determined by a single gene. Each of the genes can have multiple alleles, show incomplete dominance or co-dominance, and can be affected by the environment.

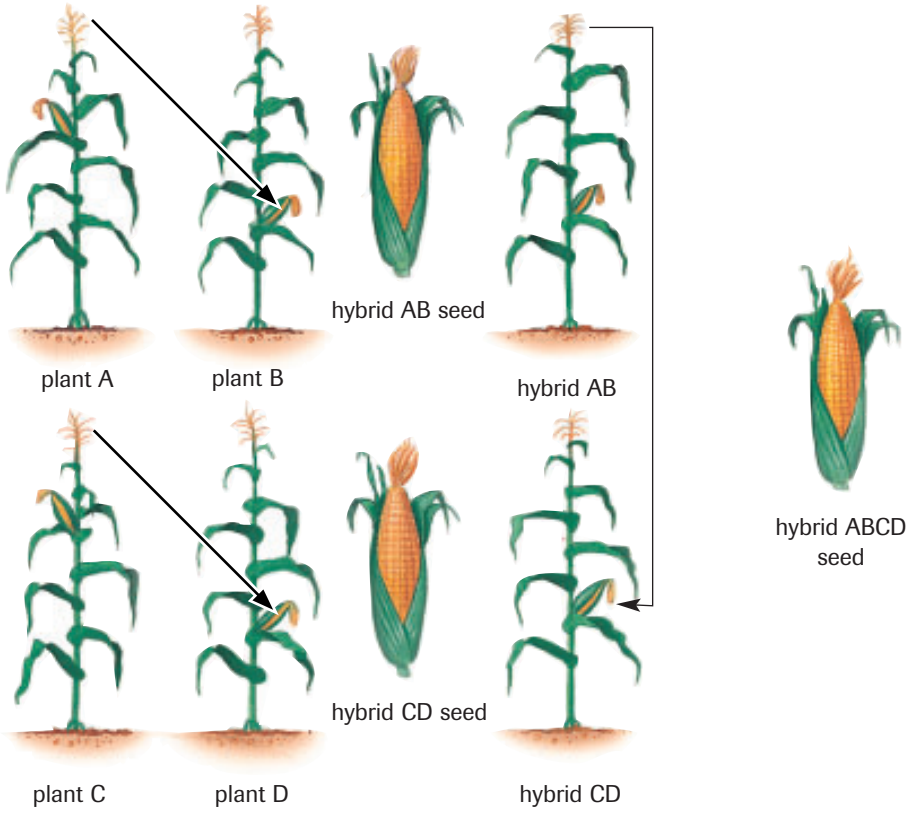

Figure 7
Hybridization can be used to produce a more vigorous strain of corn.

EXTENSION

Coat Colour in Labrador Retrievers

Coat colour variations in this breed of dog is determined by two interacting genes. In this simulation, choose a genotype for each gene and observe the phenotype.

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Examples of polygenic traits in humans include skin colour, height, and intelligence. In other animals and plants, many desirable traits, such as milk production in cows or yield in canola, are also determined by more than one gene pair. This makes breeding for these traits very difficult.

In some cases, two different genotypes interact to produce a phenotype that neither is capable of producing by itself. In other cases, one of the genes will interfere with the expression of the other, masking its effect. Genes that interfere with the expression of other genes are said to be **epistatic**.

Observed phenotypic ratios of polygenic traits vary significantly from the phenotypic ratios predicted by Punnett square analysis of non-interacting genes. Coat colour in dogs provides an examp1e of epistatic genes. As shown in the Punnet square in **Figure 8**, the allele *B* produces black coat-colour, while the recessive allele *b* produces brown coat-colour. However, a second gene also affects coat-colour. The allele *W* of this second gene prevents the formation of pigment, thereby preventing colour. The recessive allele *w* does not prevent colour. The genotype *wwBb* would be black, but the genotype *WwBb* would appear white. The *W* allele masks the effect of the *B* colour gene. In humans, the gene responsible for albinism is epistatic. This gene interferes with the expression of genes that determine pigment formation in the skin, hair, and eyes.

epistatic gene a gene that masks the expression of another gene or genes

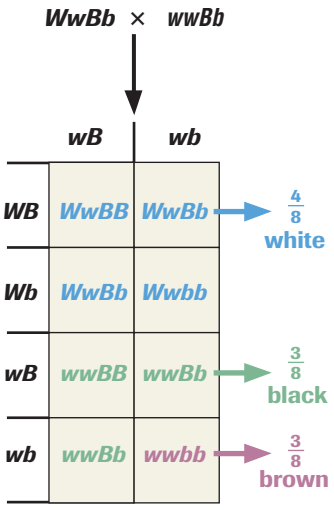

Figure 8
Punnett square of a cross between a white dog (*WwBb*) and a black dog (*wwBb*)

EXPLORE an issue

Issue Checklist

○ Issue	○ Design	● Analysis
● Resolution	● Evidence	● Evaluation

Drought-Tolerant and Salt-Tolerant Plants

Unwise agricultural practices have dramatically reduced the productivity of the world's agricultural land. By one estimate, the reduction in crop yields since 1940 is the same as if all the land in India and China had produced no crops at all. In addition, land equivalent to the area of Hungary has become so degraded that it is unable to produce any viable crop at all. Much of the problem is linked to poor irrigation techniques (**Figure 9**). When water, rich in minerals, floods the land, evaporation carries away water but leaves the minerals. Eventually, the mineral salts accumulate within the soil. creating an environment difficult for plants to survive.

Proposed Solutions from Genetics

Traditionally, plant breeders have used selective breeding to create new varieties with desirable traits. Today, molecular biologists have developed gene insertion techniques that provide breeders with a more precise tool. Using gene splicing, desired traits from one species can be introduced into a non-related species.

In 2001, articles in scientific journals reported the production of genetically modified (GM) tomatoes that can grow in soils with high salt levels. Researchers inserted a gene that enhanced the ability of cells in the tomato plants to transport excess salts into fluid storage sacs (vacuoles). The GM tomatoes can grow in soils 50 times more saline than non-GM tomatoes. The salts accumulate in the leaves, so the tomato fruit does not have a salty taste. The development of other plants capable of living in saline solutions will allow farmers to reclaim marginal land.

In related research, geneticists are looking at developing drought-tolerant plants. Several genes have been identified that enable plants to cope with arid conditions. The Rockefeller Foundation committed $50 million to support the effort to improve drought resistance for GM maize and rice.

However, as with any technology, GM drought-tolerant and salt-tolerant plants could have undesirable consequences. Some of these concerns are outlined below.

Environmental Concerns: Every year, some of the best farmland in the world is converted to urban land. This expansion of cities into farmland also reduces food production. Producing GM drought-tolerant and salt-tolerant plants that can grow on marginal land does nothing to resolve the issue of urban expansion.

GM drought-tolerant and salt-tolerant plants could lead to the conversion of deserts and saltwater marshes into agricultural land, disrupting the natural balance within these ecosystems. These ecosystems provide habitat for many species, and saltwater marshes also help filter and clean water systems.

Food Production Concerns: At present, 5 billion people inhabit Earth, and the population is projected to increase to nearly 10 billion within 50 years. Only 3.7 billion ha (hectares) of the world's 13.1 billion ha of land can be used for crop production. According to the United Nations Food and

Figure 9
Irrigation allows plants to grow in arid lands.

Agricultural committee, over the next 50 years, the amount of arable land on Earth per person will decline from 0.24 ha to about 0.12 ha, which will not be enough to feed many of the poor. Although GM crops may not be the entire answer, they may allow an increase in food production, and so deserve further study.

Geneticists' Concerns: Some geneticists worry about the consequences if GM crops hybridize with non-GM species. Traditional methods of crop breeding involve selecting particular individuals with desirable traits from within a population, thereby altering gene frequencies within a population of a single species. Newer technologies allow genes to be transferred between entirely different species. It is difficult to predict how these transferred genes will interact in a naturally reproducing population. For example, would a gene that increases drought tolerance also make a plant more susceptible to disease?

- Evaluate each of the concerns expressed.
  - (a) What assumptions lie at the basis of these divergent opinions?
  - (b) What additional information would be useful to make an informed decision about whether or not GM crops should be pursued?
- Working in a group, discuss the different viewpoints presented above.
- Still in your group, conduct additional research on the issue of developing GM drought-tolerant and salt-tolerant plants. When research is complete, discuss the question below until you reach a consensus.
  - (c) Should GM crops, resistant to drought and salinity, be funded? Do they provide at least a partial solution?
- Be prepared to debate the issue as a class. Express your opinion and provide a rationale for your view.

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SUMMARY Dihybrid Crosses

- The phenotypic ratios that Mendel observed in his dihybrid crosses provide evidence for independent assortment of chromosomes.
- The probability of inheritance of the two traits together is the same as the product of the probability of inheritance of both traits separately.

Section 18.5 Questions

1. In guinea pigs, black coat colour (*B*) is dominant to white (*b*), and short hair length (*S*) is dominant to long (*s*). Indicate the genotypes and phenotypes from the following crosses:
   (a) A guinea pig that is homozygous for black and heterozygous for short hair crossed with a white, long-haired guinea pig.
   (b) A guinea pig that is heterozygous for black and for short hair crossed with a white, long-haired guinea pig.
   (c) A guinea pig that is homozygous for black and for long hair crossed with a guinea pig that is heterozygous for black and for short hair.

2. Black coat colour (*B*) in cocker spaniels is dominant to white coat colour (*b*). Solid coat pattern (*S*) is dominant to spotted pattern (*s*). The gene for pattern arrangement is located on a different chromosome than the one for colour, and the pattern gene segregates independently of the colour gene. A male that is black with a solid pattern mates with three females. The mating with female A, which is white and solid, produces four pups: two black, solid, and two white, solid. The mating with female B, which is black and solid, produces a single pup, which is white, spotted. The mating with female C, which is white and spotted, produces four pups: one white, solid; one white, spotted; one black, solid; one black, spotted. Indicate the genotypes of the parents.

3. For human blood, the alleles for types A and B are codominant, but both are dominant over the type O allele. The Rh factor is separate from the ABO blood group and is located on a separate chromosome. The Rh+ allele is dominant to Rh−. Indicate the possible phenotypes of a child of a woman with type O, Rh− and a man with type A, Rh+.

4. Skin colour in humans is determined by more than one gene pair, whereas Rh factor in blood is controlled by one gene pair. Which would show more variability in the human population? Why?

INVESTIGATION 18.1

How Do Environmental Factors Affect Gene Expression?

Many environmental factors can affect the phenotype of a plant. Traits such as growth rate, colour, leaf size, and leaf shape can be affected by environmental factors such as light intensity, hours of darkness, wavelength of radiation, and air temperature. In this investigation, you will design an experiment to explore how one environmental factor of your choice affects the phenotype of a plant.

Report Checklist

● Purpose	● Design	● Analysis
● Problem	● Materials	● Evaluation
● Hypothesis	● Procedure	○ Synthesis
● Prediction	● Evidence	

You can find more information about designing an experiment in Appendix A1. Have your teacher check the procedure before beginning the experiment. Then, write a lab report, following the guidelines in Appendix A3.

INVESTIGATION 18.2

Genetics of Corn

Corn is one of the world's most important food crops. It has been subject to selective breeding techniques and hybridization for many years, which have resulted in vigorous, high-yielding varieties. Nearly all corn grown today is hybrid corn. Some varieties of corn are chosen for their sweet flavour while the mixed coloration of other, inedible varieties makes them popular decorations during the autumn months.

Report Checklist

○ Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	● Evidence	

Purpose

To determine the genotypes of parents by examining phenotypes of corn for two different and independent traits.

Problem

To determine the probable genotypes of the parents of the sample corn ears.

Materials

dihybrid corn ears (sample A, sample B)

Procedure

1. Obtain a sample A corn ear from your instructor (**Figure 1**). The kernels display two different traits that are located on different chromosomes.

Figure 1

(a) Indicate the two different traits.

(b) Predict the dominant phenotypes.

(c) Predict the recessive phenotypes.

INVESTIGATION 18.2 *continued*

2. Assume that the ear of corn is from the F_2 generation. The original parents were pure breeding homozygous for each of the characteristics. Assign the letters *P* and *p* to the alleles for colour, and *S* and *s* to the alleles for shape. Use the symbols *PPss* × *ppSS* for the parent generation.

(d) Indicate the phenotype of the *PPss* parent.

(e) Indicate the phenotype of the *ppSS* parent.

3. Count 100 of the kernels in sequence, and record the actual phenotypes in a table similar to **Table 1**.

Table 1 Phenotypes of the F_2 Generation

Phenotype	Number	Ratio
dominant genes for colour and shape		
dominant gene for colour, but recessive for shape		
recessive gene for colour, but dominant gene for shape		
recessive genes for colour and shape		

4. Obtain sample B. Assume that this ear was produced from a test cross. Count 100 kernels in sequence and record your results.

Analysis and Evaluation

(f) Indicate the expected genotypes and phenotypes of the F_1 generation resulting from a cross between the original parents *PPss* × *ppSS*.

(g) Use a Punnett square to show the expected genotypes and the phenotypic ratio of the F_2 generation. Compare your results with what you obtained in question 3. What factors might account for discrepancies?

(h) Assuming that sample B was produced from a test cross, indicate the phenotypic ratio of the F_1 generation.

(i) Indicate the phenotype of the unknown parent.

Synthesis

(j) Why are test crosses important to plant breeders?

(k) A dihybrid cross can produce 16 different combinations of alleles. Explain why 100 seeds were counted rather than only 16.

(l) A dominant allele *Su*, called starchy, produces smooth kernels of corn. The recessive allele *su*, called sweet, produces wrinkled kernels of corn. The dominant allele *P* produces purple kernels, while the recessive *p* allele produces yellow kernels. A corn plant with starchy, yellow kernels is cross-pollinated with a corn plant with sweet, purple kernels. One hundred kernels from the hybrid are counted, and the following results are obtained: 52 starchy, yellow kernels and 48 starchy, purple kernels. What are the genotypes of the parents and the F_1 generation?

(m) The wild ancestor of corn grew only in Central America. From this ancestor, Aboriginal peoples used selective breeding to develop different types of corn. Today, scientists continue to use technology and selective breeding methods to develop varieties of corn that can grow in a wide range of environmental conditions. As a result, corn is now grown in many places where its ancestor would not be able to survive. What are some risks associated with growing a species in a foreign environment?

EXTENSION

Comb Shape in Chickens

Two genes interact to produce comb shape in chickens. Change the genotype and see what happens to the phenotype .

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Chapter 18 SUMMARY

Outcomes

Knowledge

- describe the evidence for dominance, segregation, and the independent assortment of genes on different chromosomes, as investigated by Mendel (18.1, 18.2)
- compare ratios and probabilities of genotypes and phenotypes for dominant/recessive alleles, multiple alleles, and incompletely dominant or codominant alleles, epistatic, and pleiotropic alleles (18.2, 18.3, 18.4, 18.5)
- explain the relationship between variability and the number of genes controlling a trait (18.3)

STS

- explain that decisions regarding the application of scientific and technological development involve a variety of perspectives (18.3)

Skills

- ask questions and plan investigations by designing a plan for collecting data to demonstrate human inheritance (18.2)
- conduct investigations and gather and record data by performing an experiment to demonstrate inheritance of a trait controlled by a single pair of genes (18.5), and by designing and performing an experiment to demonstrate that an environmental factor can cause a change in the expression of genetic information in an organism (18.4)
- analyze data and apply mathematical and conceptual models by predicting, quantitatively, the probability of inheritance from monohybrid and dihybrid (18.2, 18.4); using Punnett squares to interpret patterns and trends associated with monohybrid and dihybrid patterns of inheritance (18.2, 18.4); performing, recording, and explaining predicted phenotypic ratios versus actual counts in genetic crosses to show a relationship between chance and genetic results (18.2, 18.4, 18.5); and drawing and interpreting pedigree charts from data on human single-allele and multiple-allele inheritance patterns (18.3, 18.4)
- work as members of a team and apply the skills and conventions of science (all)

Key Terms

18.1

progeny
dominant trait
recessive trait
allele
homozygous
heterozygous
genotype
phenotype
segregation

18.2

phenotypic ratio
Punnett square
genotypic ratio
test cross

18.3

pedigree chart

18.4

pleiotropic gene
wild type
mutant
incomplete dominance
codominance

18.5

dihybrid cross
selective breeding
inbreeding
polygenic trait
epistatic gene

MAKE a summary

1. Create a concept map that shows the principles of inheritance of traits. Label the sketch with as many of the key terms as possible.
2. Revisit your answers to the Starting Points questions at the start of the chapter. Would you answer the questions differently now? Why?

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The following components are available on the Nelson Web site. Follow the links for *Nelson Biology Alberta 20–30*.

- an interactive Self Quiz for Chapter 18
- additional Diploma Exam-style Review Questions
- Illustrated Glossary
- additional IB-related material

There is more information on the Web site wherever you see the Go icon in the chapter.

EXTENSION

CBC radioONE QUIRKS & QUARKS

Spawning Trouble

Dr. Daniel Heath, (University of Windsor) has discovered that the eggs of captive-bred salmon are getting smaller each year. The lack of selective pressure on the eggs in a hatchery may be the cause, since more small fish are surviving than would be if the eggs developed in the wild. Dr. Heath is concerned this will lead to health problems in the wild population, and if this may also be a general problem with captive breeding programs for other animals, including endangered species.

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Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

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DO NOT WRITE IN THIS TEXTBOOK.

Part 1

Use the following information to answer questions 1 and 2.

Long stems are dominant over short stems for pea plants. A heterozygous long-stem plant is crossed with a short-stem plant.

1. Determine and identify the genotypic ratio of the F_1 progeny from the cross.
   A. 50 % *Ss* and 50 % *ss*
   B. 75 % *SS* and 25 % *Ss*
   C. 75 % *Ss* and 25 % *ss*
   D. 100 % *Ss*

2. Determine and identify the phenotypic ratios of the F_1 progeny of the cross.
   A. 75 % long stem and 25 % short stem
   B. 50 % long stem and 50 % short stem
   C. 75 % short stem and 25 % long stem
   D. 100 % long stem

Use the following information to answer questions 3 to 5.

The pedigree chart in **Figure 1** shows the transmission of blood types in a family.

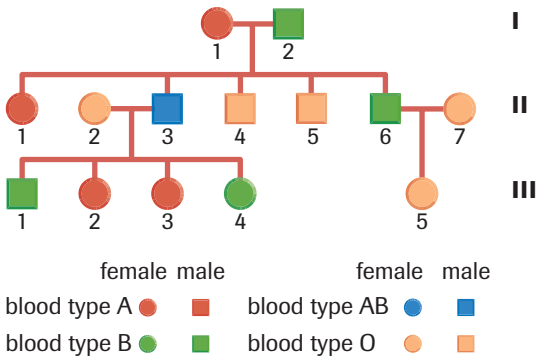


Figure 1

3. Indicate the genotypes for individuals 1 and 2, generation I.
   A. I^Ai and I^Bi
   B. I^AI^A and I^BI^B
   C. I^Ai and I^BI^B
   D. I^AI^B and I^Bi

4. NR Predict the chance of parents 1 and 2 from generation I having a child with blood type AB. (Record your answer in decimal form.)

5. NR If individuals 6 and 7 had another child, calculate the probability that the child would have blood type O. (Record your answer in decimal form.)

Use the following information to answer questions 6 and 7.

In cattle, the polled trait (hornless) is dominant to the horned condition. A single bull mates with three different cows and produces offspring as shown in **Figure 2**.

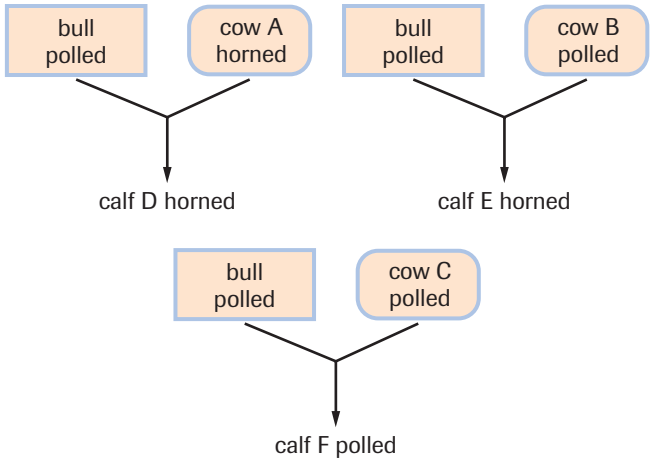


Figure 2

6. Identify the respective genotypes for the bull, cow A, and cow B.
   A. bull = *Pp*, cow A = *pp*, cow B = *Pp*
   B. bull = *PP*, cow A = *pp*, cow B = *Pp*
   C. bull = *Pp*, cow A = *pp*, cow B = *pp*
   D. bull = *PP*, cow A = *Pp*, cow B = *Pp*

7. Identify which of the cattle could have two possible genotypes.
   A. cow C and calf F
   B. cow B and calf E
   C. cow A and calf D
   D. bull and calf D

Part 2

8. **Explain** the advantages and limitations of using blood typing by the courts to prove paternity.

9. Cystic fibrosis is regulated by a recessive allele, *c*. **Explain** how two parents without this condition can produce a child with cystic fibrosis.

10. In horses, the trotter trait is dominant to the pacer trait. A male, described as a trotter, mates with three different females. Each female produces a foal. The first female, a pacer, gives birth to a foal that is a pacer. The second female, also a pacer, gives birth to a foal that is a trotter. The third female, a trotter, gives birth to a foal that is a pacer. **Determine** the genotypes of the male, all three females, and the three foals sired. Designate the trotter allele as *T* and the pacer allele as *t*.

11. For ABO blood groups in humans, the A and B genes are codominant, but both A and B are dominant over type O.
(a) **Identify** the possible blood types in the children of a man with blood type O and a woman with blood type AB.
(b) Could a woman with blood type AB ever produce a child with blood type AB? Could she ever have a child with blood type O? **Explain** your answer.

12. Some cats have six toes, a condition determined by a dominant allele. **Sketch** a pedigree chart showing the mating of a male cat with six toes to a normal female. Assume the following:
- The male cat with six toes had a normal mother.
- The cats produce six offspring (four females and two males). Two of the female offspring and one of the male offspring have six toes.
- One of the six-toed female offspring mates with a six-toed male from different parents. Four female offspring are produced, and three of them have six toes.

13. In shorthorn cattle, the mating of a red bull and a white cow produces a calf that is described as roan. Roan animals have intermingled red and white hair. After many matings between roan bulls and roan cows, the following phenotypic ratio was observed in the offspring: one red, two roan, one white. Does this ratio indicate codominance or multiple alleles? **Explain** your answer.

Use the following information to answer questions 14 to 16.

Thalassemia is a serious human genetic disorder which causes severe anemia. The homozygous condition (T^mT^m) leads to severe anemia. People with thalassemia die before sexual maturity. The heterozygous condition (T^mT^n) causes a less serious form of anemia. The genotype T^nT^n causes no symptoms of the disease.

DE **Predict** all the possible genotypes of the offspring of a male with the genotype T^mT^n and a woman of the same genotype.

DE **Predict** all the possible phenotypes of the offspring of a man with the genotype T^mT^n and a woman of the same genotype.

DE Would it ever be possible for offspring to be produced from two individuals with the genotypes T^mT^m and T^mT^n respectively? **Explain** your answer.

Use the following information to answer questions 17 and 18.

Baldness is an autosomal trait, but it is influenced by sex. Baldness (*HB*) is dominant in males but recessive in females. The normal gene (*Hn*) is dominant in females, but recessive in males.

17. DE **Explain** how a bald offspring can be produced from the mating of a normal female and a normal male.

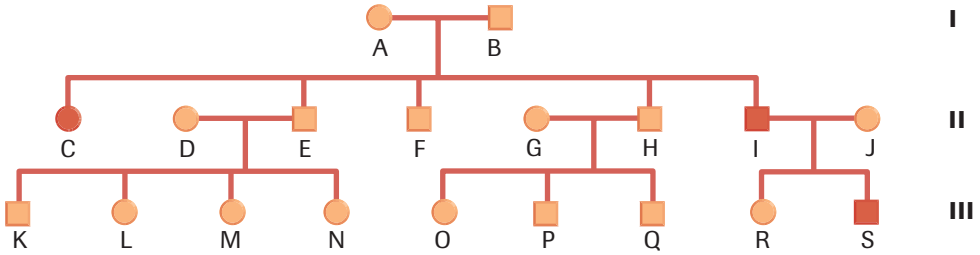
18. DE Could normal parents ever produce a bald girl? **Explain** your answer.

19. The ability to curl your tongue up on the sides (*T*) is dominant to not being able to roll your tongue (*t*).
(a) A woman who can roll her tongue marries a man who cannot. Their first child has his father's phenotype. **Predict** the genotypes of the mother, father, and child.
(b) **Determine** the probability that their second child will not be able to roll her or his tongue.

20. Phenylketonuria (PKU) is an inherited disease caused by the lack of the enzyme needed to metabolize the amino acid phenylalanine. If untreated, PKU builds up in the brain and causes mental retardation. PKU is determined by a recessive allele. A woman and her husband are both carriers of PKU. **Determine** the probability of
(a) their first child having PKU.
(b) both of their first two children having PKU.

21. DE Amniocentesis is a common prenatal procedure, used to obtain cells to test for genetic abnormalities such as cystic fibrosis. The test is usually carried out in the 15th to 18th week of pregnancy when a woman has an increased risk of having children with genetic abnormalities. A woman with cystic fibrosis in her family history (**Figure 3**, next page) is carrying a child. Her husband's lineage also is linked to cystic fibrosis. Cystic fibrosis is caused by a recessive allele found on chromosome 7. Write a unified response addressing the following aspects of performing amniocentesis in the case of father K and mother O.
- Like all procedures that enter the body, some risk, although small, is associated with amniocentesis. On the basis of the information provided, would you recommend an amniocentesis be done for mother O and father K? **Explain** your reasons.
- Would you recommend the procedure if father K had married mother O's cousin, woman J? **Explain** your reasons.
- Should amniocentesis be performed even if there is no strong evidence suggesting genetic problems? **Explain** your reasons.
- Should this pedigree be made public? **Identify** both pros and cons before coming to a conclusion.

Father K's Family Tree



Mother O's Family Tree

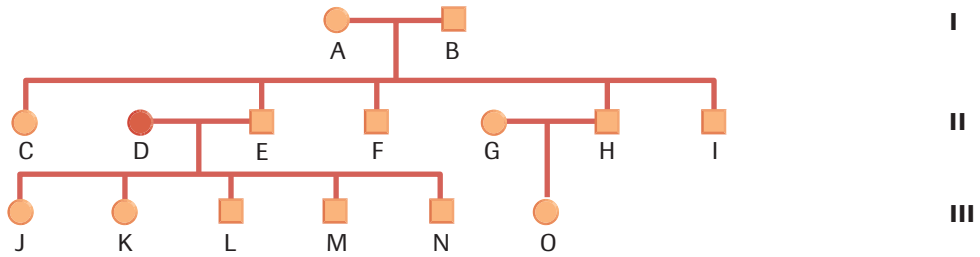


Figure 3

22. In Canada, it is illegal to marry your immediate relatives. Using the principles of genetics, **explain** why inbreeding of humans is discouraged.

Use the following information to answer questions 23 to 26.

When paper impregnated with the bitter chemical phenylthiocarbamide (PTC) is placed on the tongue, about 70 % of people can taste the chemical. The ability to taste PTC is determined by a dominant taster allele (*T*). Those who cannot taste PTC are homozygous for the recessive alleles (*t*). A second gene on another chromosome determines skin pigmentation. Allele (*A*) is dominant, and determines normal pigmentation. People who are homozygous for the recessive allele (*a*) will be albino. A normally pigmented woman who cannot taste PTC has a father who is an albino and a PTC taster. She marries a normally pigmented man who is homozygous for the dominant (A) allele for pigmentation. The man can taste PTC, but his mother cannot.

23. **Predict** all the possible genotypes for these two traits for children by this couple. DE

24. **Determine** the probability that a child from this couple will not be able to taste PTC. DE

25. **Determine** the probability that a child from this couple will be albino? DE

26. **Determine** the probability that a child from this couple will be able to taste PTC and be albino. DE

Use the following information to answer questions 27 to 29.

In a specific variety of soybeans, the allele for seeds containing a high oil-content (*H*) is dominant to the allele for low oil-content (*h*). A gene located on another chromosome determines the number of seeds in a pod. Through crossing experiments, it was determined that the allele that determines four seeds per pod (*E*) is dominant to the allele that determines two seeds per pod (*ee*). A plant breeder crosses two soybean plants of this variety, both of which have high oil-content and four seeds per pod. The phenotypes of the F_1 generation and their ratios are shown in **Table 1**.

Table 1 Phenotypes of the F_1 Generation

Phenotype	Ratio
high oil-content—four seeds per pod	9
high oil-content—two seeds per pod	3
low oil-content—four seeds per pod	3
low oil-content—two seeds per pod	1

27. **Predict** the genotypes of the parent plants. DE

28. The plant breeder crosses two individuals from the F_1 generation that have high oil-content and four seeds per pod. If all the members of the F_2 generation all have high oil-content and four seeds per pod, **predict** the genotypes of the two F_1 parent plants chosen by the breeder. DE

29. The breeder wants to confirm the genotype of the two F_1 parent plants using a cross. What genotype should the plant she crosses the F_1 parent plants have? **Explain**. DE